Adult E-Poster

CONCLUSION
Swyer syndrome may mimic Turner syndrome in cases of primary amenorrhea with short stature. Accurate diagnosis requires comprehensive hormonal, imaging and genetic evaluation beyond clinical phenotype alone.

EP_A088

UNRAVELLING THE MYSTERY: A CASE OF ATYPICAL DIABETES WITH HEPATIC AND RENAL CLUES TO HNF1B DEFICIENCY

https://doi.org/10.15605/jafes.040.S1.096

Asma' Mohd Nazlee, Florence Tan Hui Sieng, Chan Pei Lin
Endocrinology Unit of Medical Department, Sarawak General Hospital, Malaysia

INTRODUCTION/BACKGROUND
Hepatocyte nuclear factor 1β (HNF1B) deficiency associated with MODY-5 is increasingly recognised as a multifaceted syndrome with diverse manifestations. We present a suspected case initially misdiagnosed as type 1 diabetes with autoimmune hepatitis.

CASE
A 14-year-old male with learning disability was admitted for insulin initiation when he presented with osmotic symptoms with hyperglycaemia and ketonuria. He reported no family history of diabetes. HbA1c was 18.5% and LFTs were deranged (AST 74, ALT 209 and ALP 451 IU/L). He has some dysmorphic facial features. Despite good glycemic control on intensive insulin therapy, his liver enzymes remained elevated (8-17x ULN) with normal ferritin, ceruloplasmin and viral panel. Abdominal ultrasound showed normal liver and spleen but detected bilateral medullary nephrocalcinosis. The liver biopsy showed mild periportal hepatitis. He was treated for autoimmune hepatitis with prednisolone and azathioprine. Subsequent investigations revealed negative diabetes (anti-GAD, ICA, IA2), hepatic (ANA, smooth muscle, LC1, LKM and mitochondrial) autoantibodies and normal serum immunoglobulins. The absence of diabetes-related autoantibodies, coupled with multisystem involvement (pancreas, liver, kidney, neurocognitive and dysmorphism), raised the suspicion of HNF1B mutation. Although genetic confirmation was not feasible, further investigation with elevated C peptide (1652 pmol/L) and persistent hypomagnesemia (0.4 to 0.55 mmol/L) further substantiated this hypothesis. Immunotherapy was withheld. He remained well with fluctuating liver function on follow-up 5 years since the initial presentation.

CONCLUSION
This case underscores the diagnostic complexity of HNF1B deficiency, a rare monogenic diabetes subtype accounting for ~6% of MODY. Despite an autosomal dominant inheritance pattern, de-novo mutation accounts for 50% of cases. Lack of family history does not preclude the diagnosis. Diagnostic clues include multisystem involvement, which is rarely found in other MODY subtypes. Hypomagnesemia is another common feature. Early recognition is essential for individualised management, avoidance of mismanagement, monitoring for other organ involvement or complications and genetic counselling.

EP_A089

TWIN-TWIN TRANSFUSION SYNDROME ASSOCIATED MATERNAL HYPERTHYROIDISM

https://doi.org/10.15605/jafes.040.S1.097

Tean Chooi Fun and Ijaz Binti Hallaj Rahmatullah
Hospital Raja Permaisuri Bainun, Perak, Malaysia

INTRODUCTION/BACKGROUND
Pregnancies complicated by twin-twin transfusion syndrome (TTTS) are associated with elevated human chorionic gonadotropin (hCG) compared to uncomplicated twin pregnancies. Studies have shown a positive correlation between hCG and free thyroxine (FT4) in TTTS, thereby increasing the risk of maternal hyperthyroidism. This case report describes a twin pregnancy complicated by TTTS, where maternal hyperthyroidism developed prior to fetoscopic laser ablation (FLA).

CASE
We present a 36-year-old female with a twin pregnancy complicated by TTTS. She was diagnosed with gestational transient thyrotoxicosis (GTT) at 10 weeks of gestation with thyroid stimulating hormone (TSH) of 0.01 mIU/L, FT4 of 24.8 pmol/L and triiodothyronine (T3) of 3.8pmol/L. She had negative thyroid-stimulating hormone receptor antibodies and a normal neck ultrasound. Clinically, she has no goitre or thyroid eye disease. At 15 weeks of gestation, her FT4 decreased to 14.3 pmol/L while TSH remained suppressed. She did not receive any anti-thyroid drugs (ATDs) during the first trimester. She was admitted at 22 weeks of age of gestation for FLA due to TTTS stage 1. Upon admission, she complained of palpitations, and the cardiac monitor showed sinus tachycardia with a heart rate of 123 bpm. Her TSH was <0.008 mIU/L, FT4 was increased to 21 pmol/L and hCG of >225,000U/L. Due to hyperthyroid symptoms, she was treated with carbimazole and beta-blocker prior to FLA. Her carbimazole dose was reduced at 25 weeks of gestation as FT4 dropped to 13.2 pmol/L. It was then